22q11.2 duplication

22q11.2 duplication is a condition caused by an extra copy of a small piece of chromosome 22. The duplication occurs near the middle of the chromosome at a location designated q11.2.

The features of this condition vary widely, even among members of the same family. Affected individuals may have developmental delay, intellectual disability, slow growth leading to short stature, and weak muscle tone (hypotonia). Many people with the duplication have no apparent physical or intellectual disabilities.

Frequency

The prevalence of the 22q11.2 duplication in the general population is difficult to determine. Because many individuals with this duplication have no associated symptoms, their duplication may never be detected.

Most people tested for the 22q11.2 duplication have come to medical attention as a result of developmental delay or other problems affecting themselves or a family member. In one study, about 1 in 700 people tested for these reasons had the 22q11.2 duplication. Overall, more than 60 individuals with the duplication have been identified.

Genetic Changes

People with 22q11.2 duplication have an extra copy of some genetic material at position q11.2 on chromosome 22. In most cases, this extra genetic material consists of a sequence of about 3 million DNA building blocks (base pairs), also written as 3 megabases (Mb).

The 3 Mb duplicated region contains 30 to 40 genes. For many of these genes, little is known about their function. A small percentage of affected individuals have a shorter duplication in the same region. Researchers are working to determine which duplicated genes may contribute to the developmental delay and other problems that sometimes affect people with this condition.

Inheritance Pattern

The inheritance of 22q11.2 duplication is considered autosomal dominant because the duplication affects one of the two copies of chromosome 22 in each cell. About 70 percent of affected individuals inherit the duplication from a parent. In other cases, the duplication is not inherited and instead occurs as a random event during the formation of reproductive cells (eggs and sperm) or in early fetal development. These affected

people typically have no history of the disorder in their family, although they can pass the duplication to their children.

Other Names for This Condition

- chromosome 22q11.2 duplication syndrome
- chromosome 22q11.2 microduplication syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: 22q11.2 duplication syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2675369/

Other Diagnosis and Management Resources

- GeneReview: 22q11.2 Duplication
  https://www.ncbi.nlm.nih.gov/books/NBK3823

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Developmental Disabilities
  https://medlineplus.gov/developmentaldisabilities.html

Genetic and Rare Diseases Information Center

- 22q11.2 duplication syndrome
  https://rarediseases.info.nih.gov/diseases/10557/22q112-duplication-syndrome

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities
  https://www.genome.gov/11508982/

Educational Resources

- Centers for Disease Control and Prevention: Intellectual Disability
  https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/IntellectualDisability.pdf
- Disease InfoSearch: 22q11.2 Duplication Syndrome
  http://www.diseaseinfosearch.org/22q11.2+Duplication+Syndrome/21
- MalaCards: chromosome 22q11.2 microduplication syndrome
  http://www.malacards.org/card/chromosome_22q112_microduplication_syndrome
- March of Dimes: Chromosomal Conditions
  http://www.marchofdimes.org/baby/chromosomal-conditions.aspx

Patient Support and Advocacy Resources

- Chromosome 22 Central
  http://www.c22c.org/about.htm
- Chromosome Disorder Outreach
  http://chromodisorder.org/
- Unique: Rare Chromosome Disorder Support Group
  http://www.rarechromo.org/
- University of Kansas: Support Groups for Chromosomal Disorders
  http://www.kumc.edu/gec/support/chromoso.html

GeneReviews

- 22q11.2 Duplication
  https://www.ncbi.nlm.nih.gov/books/NBK3823

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%2822q11.2+duplication%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CHROMOSOME 22q11.2 DUPLICATION SYNDROME
  http://omim.org/entry/608363

Sources for This Summary

- Courtens W, Schramme I, Laridon A. Microduplication 22q11.2: a benign polymorphism or a syndrome with a very large clinical variability and reduced penetrance?--Report of two families. Am J Med Genet A. 2008 Mar 15;146A(6):758-63. doi: 10.1002/ajmg.a.31910.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18260141
- GeneReview: 22q11.2 Duplication
  https://www.ncbi.nlm.nih.gov/books/NBK3823
- Mukaddes NM, Herguner S. Autistic disorder and 22q11.2 duplication. World J Biol Psychiatry. 2007;8(2):127-30.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17455106
- Ou Z, Berg JS, Yonath H, Enciso VB, Miller DT, Picker J, Lenzi T, Keegan CE, Sutton VR, Belmont J, Chinault AC, Lupski JR, Cheung SW, Roeder E, Patel A. Microduplications of 22q11.2 are frequently inherited and are associated with variable phenotypes. Genet Med. 2008 Apr;10(4): 267-77. doi: 10.1097/GIM.0b013e31816b64c2.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18414210
- Portnoï MF. Microduplication 22q11.2: a new chromosomal syndrome. Eur J Med Genet. 2009 Mar-Jun;52(2-3):88-93. doi: 10.1016/j.ejmg.2009.02.008. Epub 2009 Feb 28. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19254783
- Ramelli GP, Silacci C, Ferrarini A, Cattaneo C, Visconti P, Pescia G. Microduplication 22q11.2 in a child with autism spectrum disorder: clinical and genetic study. Dev Med Child Neurol. 2008 Dec; 50(12):953-5. doi: 10.1111/j.1469-8749.2008.03048.x.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19046189
- Torres-Juan L, Rosell J, Morla M, Vidal-Pou C, García-Algas F, de la Fuente MA, Juan M, Tubau A, Bachiller D, Bernues M, Perez-Granero A, Govea N, Busquets X, Heine-Suñer D. Mutations in TBX1 genocopy the 22q11.2 deletion and duplication syndromes: a new susceptibility factor for mental retardation. Eur J Hum Genet. 2007 Jun;15(6):658-63. Epub 2007 Mar 21.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17377518
- Wentzel C, Fernström M, Ohrner Y, Annerén G, Thuresson AC. Clinical variability of the 22q11.2 duplication syndrome. Eur J Med Genet. 2008 Nov-Dec;51(6):501-10. doi: 10.1016/j.ejmg.2008.07.005. Epub 2008 Jul 29.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18707033
- Yu S, Cox K, Friend K, Smith S, Buchheim R, Bain S, Liebelt J, Thompson E, Bratkovic D. Familial 22q11.2 duplication: a three-generation family with a 3-Mb duplication and a familial 1.5-Mb duplication. Clin Genet. 2008 Feb;73(2):160-4. Epub 2007 Dec 12.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18076674

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